CROSS PARTY GROUP ON RARE, GENETIC AND UNDIAGNOSED CONDITIONS

Date: 16 June 2021

Venue: Zoom meeting

MINUTES

	Welcome and Introductions Bob Doris MSP welcomed all in attendance and advised that this was the first meeting to re-establish the CPG . Following today's meeting, paperwork will be submitted to the Standards, Procedures and Public Appointments Committee. Once the paperwork has been accepted, the CPG will be constituted and members informed. ACTION: Natalie to inform CPG members once paperwork has been accepted and circulate the date for next meeting. **Overview of work of CPG in last session** Natalie Frankish provided a brief overview of the work of the CPG in the last Parliament, including a summary of the CPG's Report 'Improving Care for Rare Conditions in Scotland'. Bob Doris MSP provided some reflections on the work of the CPG and noted the importance of the input from patient group members in shaping the work plan of the group. Bob thanked members for sharing their experiences and for the valuable suggestions and recommendations that had informed the CPG report. A motion to re-establish CPG was raised and accepted. It was agreed that no subscription charge would be required in recognition of the small size and limited resource of many patient organisations. Genetic Alliance UK will provide the Secretariat for the CPG. It was noted that, as in previous years, only patient organisations be considered as non-MSP members – this means their membership is listed on the Scottish Parliament website and they have voting rights. All other attendees (for example individuals, health care professionals or academics) are welcome to attend meetings and contribute in an observing capacity. This ensures that the CPG has an opportunity to hear from all stakeholders, but the CPG remains firmly a forum for patients and their representatives.

Role of the CPG

The CPG should expect to play an important role in the development and implementation of the new UK Framework for Rare Diseases and the Scottish Implementation Action Plan.

This meeting will provide an introduction to the key priorities of the UK Framework in the Scottish Context. A panel of speakers were invited.

Implementing the UK Rare Disease Framework

- **Introduction –Scottish Government Clinical Priorities Team**
  Sarah Ogilvie provided an overview of the UK Rare Diseases Framework and the Scottish Government's plans for developing a Rare Diseases Action Plan for Scotland. Sarah noted that the experiences of people living with rare conditions in Scotland would inform the plan and that a patient advisory group would be established in due course. Sarah also noted that the CPG's views on the Plan would be sought and the CPG's recent report would be considered by the Rare Disease Implementation Group.
- **Diagnosis (Professor Zosia Miedzybrodzka, Service Clinical Director of Genetics, NHS Grampian)**
  Professor Miedzybrodzka provided an overview on how genomic services are delivered and funded in Scotland, including information on the type of the testing offered, it's value to patients and details of the bridge funding required to support the service.
- **Raising awareness with health professionals (Dr Martina Rodie, Office for Rare Conditions, Glasgow)**
  Dr Rodie described the role of the Office for Rare Conditions and it's work to raise awareness of rare conditions. Dr Rodie described the programme of engagement events held for clinicians and other health professionals in Scotland and the important role of providing information, support and training to health professionals.
- **Care Coordination and Access to specialist care, treatment and drugs (Dr Catherine McWilliam, Scottish Muscle Network)**
  Dr McWilliam provided a presentation on Spinal Muscular Atrophy, with details of a new treatment available for the condition. Dr McWilliam noted that the limitations of the Newborn Screening Programme meant that there were delays in identifying children who could benefit from the treatment, and this was often detrimental to the patient outcomes for the child. Expansion of the Newborn Screening Programme in Scotland would help to address this.

ACTION: Dr McWilliam to write to Bob Doris MSP with further information.

Discussion of CPG Work Plan 2021-2022

It was agreed that the CPG could play an important role in informing the development of the Scottish Government's Rare Diseases Action Plan. There was support for a programme of meetings focusing on each of the

	Framework themes (diagnosis, raising Awareness with health professionals, care coordination and access to specialist care, treatment and drugs). It was also noted that the CPG should monitor the impact of Covid-19, and where possible inform the re-mobilisation of services. **ACTION**: Natalie Frankish to circulate details of next meeting
	Meeting attendees Bob Doris MSP Paul McLennan MSP Natalie Frankish – Genetic Alliance UK Professor Zosia Miedzybrodzka – University of Aberdeen Dr Catherine McWilliam – Scottish Muscle Network Dr Martina Rodie – Office for Rare Conditions (Glasgow) Claire Fyvie Kate Murdoch Rae McNairney Michelle Conway Harriette Campbell Mike Cain (HSP Support) Katherine Behl (AHC UK) John Wallace Amy Comrie Rachael Scott Arlene Smyth (Turner Syndrome Support Society) Janice Connelly Dan Farthing (Haemophilia Scotland) Salena Begley (Family Fund) Tony Thornburn (Behcet's UK) Catherine O'Hara (Behcet's UK) Michelle Erskine (The Aarskog Foundation) Lynne Hocking-Mennie Andrew Deans